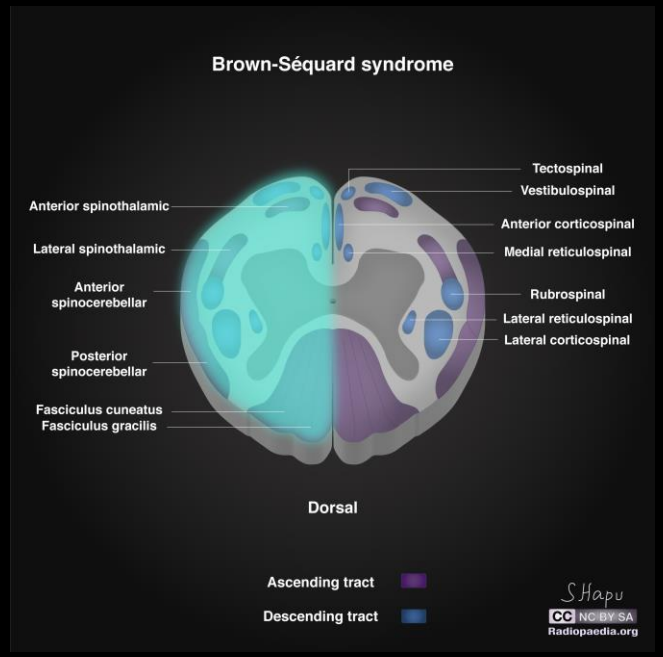
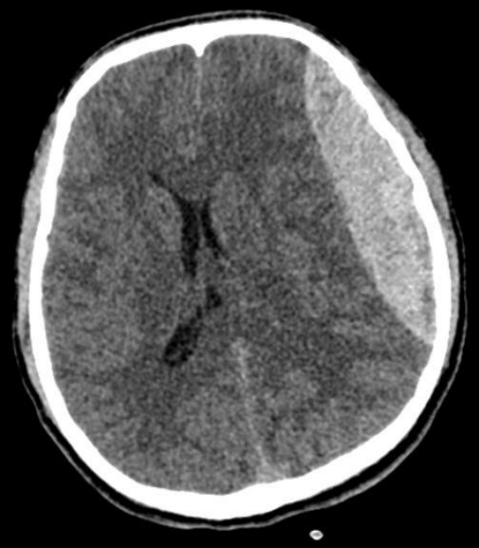
# High Yield Neurology Review

By Evan, Mike




Case courtesy of Sachi Hapugoda, Radiopaedia.org, rID: 62852



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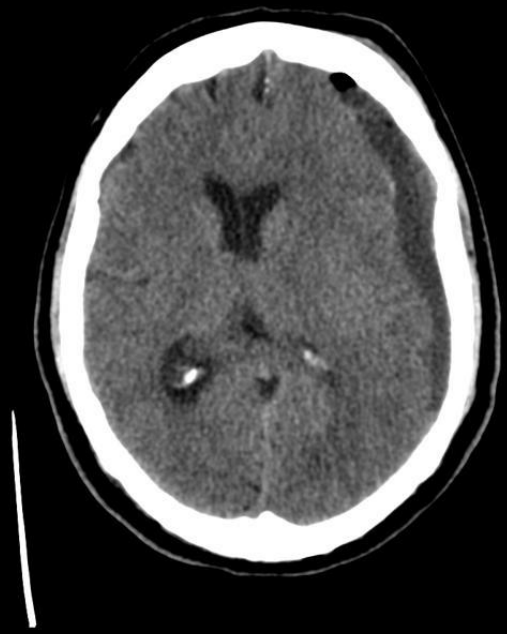

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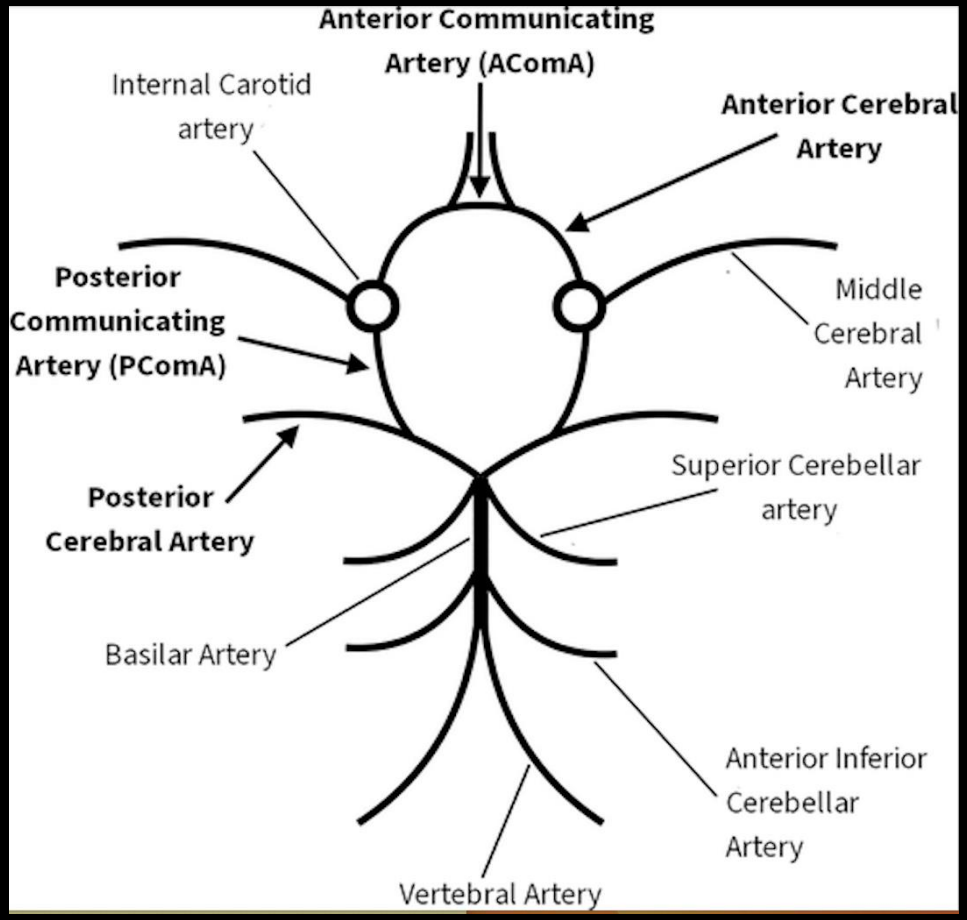
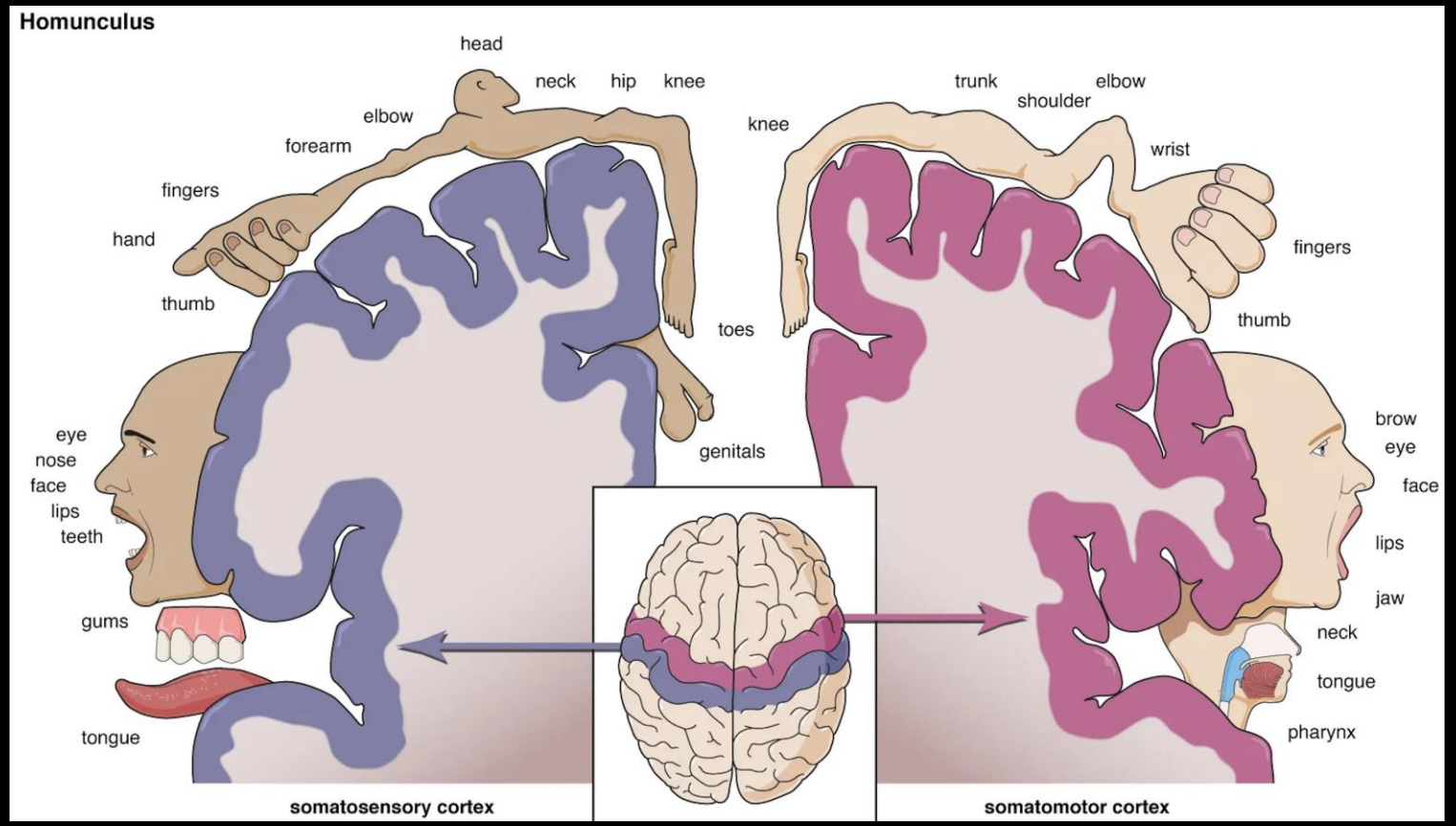
# Clinical Neuroanatomy




Anatomical variations of the circle of Willis and their prevalence…
*Clinical Anatomy*. 34:7; 978-990




Britannica

# Clinical Neuroanatomy: Spinal Cord

## Ascending columns

Pressure, vibration, fine touch, two-point discrimination conscious proprioception?

Dorsal

Pain, temperature, crude touch?

Spinothalamic tract

Unconscious proprioception?

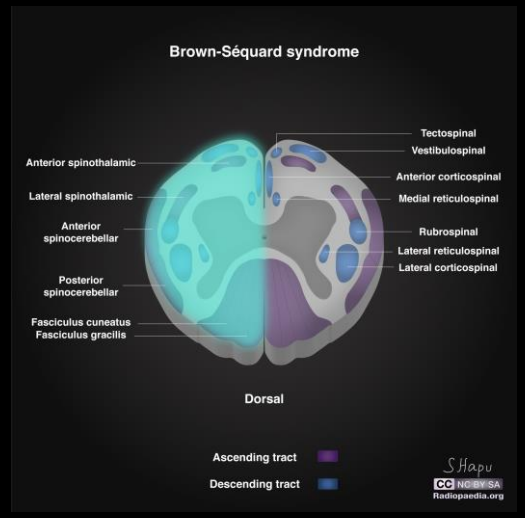
Posterior spinocerebellar

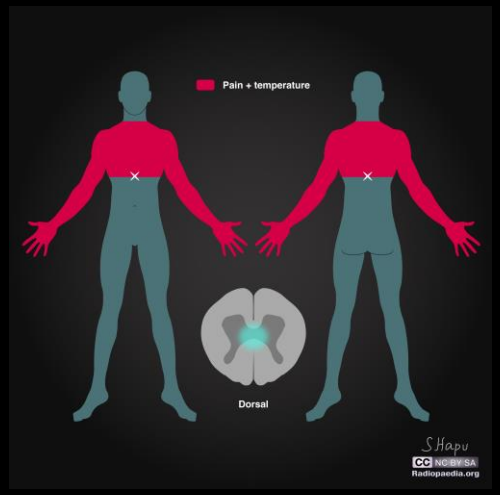
## Descending columns

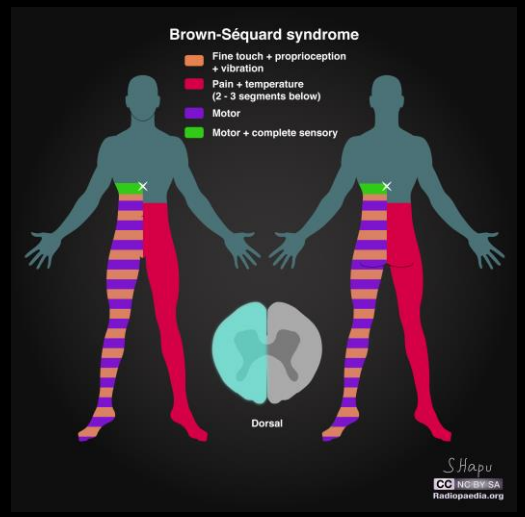
Voluntary motor?

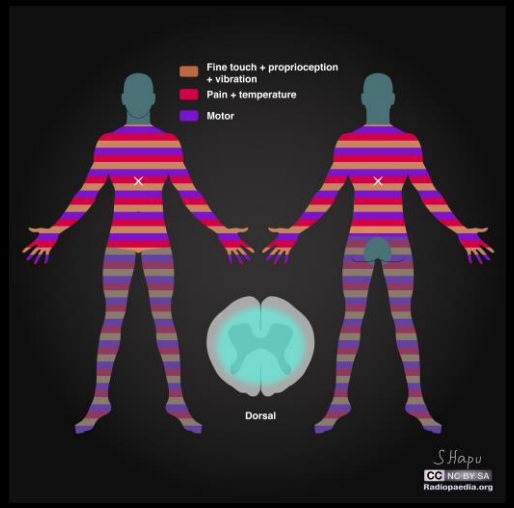
Anterior/lateral corticospinal tract

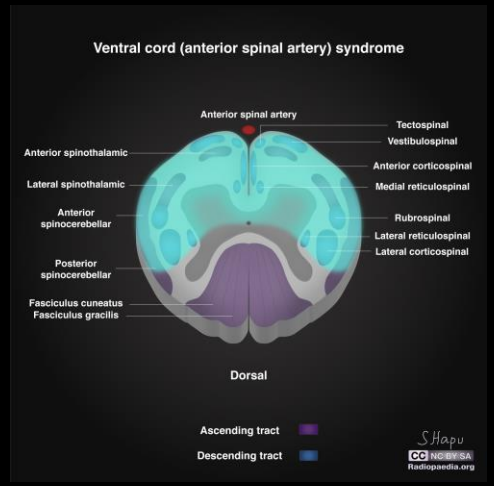
# Clinical Neuroanatomy: Spinal Cord




Case courtesy of Sachi Hapugoda, Radiopaedia.org, rID: 62852

**32 M stab wound to T10 on R with ipsilateral hemiparesis/loss of vibration/proprioception below lesion and contralateral loss of pain/temperature 1-2 levels below?**

Brown-Sequard

**Bilateral LE weakness and loss of pain/temperature sensation w/ preserved proprioception/vibratory after MVC?**

Anterior cord syndrome

Pain/temperature loss/weakness in UEs?

Central cord syndrome

Main 2 etiologies?

Syrinx (chronic), neck hyperextension (acute trauma)

**Ataxic gait, paresthesias, impaired position/vibration sense and anemia?**

Subacute combined degeneration (vitamin B12 deficiency)

Pathophys?

Demyelination of dorsal columns, lateral corticospinal tracts, and spinocerebellar tracts

**Symmetric LE weakness, hyperreflexia, bowel/bladder dysfunction?**

Conus medullaris syndrome

**Severe radicular leg pain, asymmetric weakness, hyporeflexia, and saddle anesthesia?**

Cauda equina syndrome

Etiologies?

Disc herniation, epidural abscess, trauma, metastatic cancer

Best next step?

Emergent MRI, surgical evaluation

Tx?

Depends on underlying etiology

- Decompressive surgery for disk herniation
- Epidural abscess: IV abx; Mets: IV steroids/surgery/radiation

# 28 yo F with throbbing, unilateral HA. Pain worse with menstruation and minimally relieved with Tylenol and lying in a dark room

Most likely dx?

Migraine HA

Triggers?

Certain foods (red wine, cheese), fasting, stress, menses, OCPs, bright lights, disruption in sleep pattern

Diagnosis?

Clinical

Tx for acute?

Abortive therapy: OTC NSAIDs, triptans, metoclopramide, ergots

Prophylaxis?

Anticonvulsants (valproate, topiramate), TCAs, propranolol (1st line in pregnancy)

# 25 yo M smoker with periodic stabbing right eye pain

Most likely dx?

Cluster headache

Tx for acute?

High-flow 100% O2

Prophylaxis?

Verapamil

32 F with BMI of 40 c/o bilateral HA worse in the morning and with Valsalva?

Idiopathic intracranial HTN

Non-pharmacologic tx? Best pharmacologic tx?

Address risk factors/comorbidities, weight loss

Acetazolamide

# 44 yo M c/o sudden severe HA, N/V, photophobia

HA red flag signs?

Sudden and severe onset, “worst headache of my life”, age >65 or <10, focal neurologic deficits, papilledema, in the setting of head trauma

Most likely dx?

Subarachnoid hemorrhage

Next best step?

Non-contrast head CT

If negative but still high suspicion?

Lumbar puncture -> xanthochromia

Tx?

Reverse anticoagulation, target SBP < 160 mmHg, ICP management (elevate head of bed, etc), neurosurgery

Complications?

Vasospasm (3-10 days), recurrent bleeding (first 24 hours), hydrocephalus, seizure, SIADH

Vasospasm prevention?

Nimodipine

# 44 y/o M with shock like stabbing pain along CN V distribution?

Trigeminal neuralgia

Pathophys?

Neurovascular compression of trigeminal nerve root

If bilateral?

Think multiple sclerosis

Tx?

Carbamazepine, oxcarbazepine

**65 yo M with weight loss, tenderness over temple, pain with chewing**

Giant cell arteritis

Diagnosis?

Inflammatory markers (ESR, CRP) very elevated (ESR>100)

Temporal artery biopsy

Tx?

High dose glucocorticoids to lower risk of permanent vision loss

# 60 yo M smoker, HTN with transient speech difficulty and right arm/leg weakness

Most likely dx?

TIA

Next best step?

Neuroimaging, EKG (possibly Echo)

Tx?

Reduce stroke risk (antithrombotic therapy, treat underlying etiology, ASCVD prevention)

Same patient but symptoms persist

Acute stroke (80% ischemic, 20% hemorrhagic)

Common etiologies?

Atherosclerosis, lacunar (cx HTN, HLD, DM), cardiac emboli, hypercoagulable states, sickle cell, dissection

Best next step?

Head CT without contrast

Further workup?

To determine cause: carotid US, EKG, echo, neuroimaging

tPA eligibility: CBC, PT/PTT, cardiac enzymes, troponin, BUN, Cr

Tx?

Thrombolytics if <3-4.5 hours since onset with no bleeding or absolute contraindications

Consider thrombectomy for large vessel occlusion within 24 hrs

Prevention/long-term management?

Treat HTN, DM, statin therapy, ASA/clopidogrel, anticoagulation for cardioembolic, carotid endarterectomy if stenosis > 70% (>80% if asymptomatic)

# 55 M smoker, HTN *worsening* focal neurologic deficits and a HA, N/V

Most likely dx?

Intracerebral hemorrhage

Most common cause?

HTN, amyloid angiopathy (in elderly)

55 M head trauma with acute LOC then lucid followed by gradual LOC

Epidural hematoma

Pathophys?

Tear of middle meningeal artery -> blood accumulation between skull and dura mater

Non-con CT findings?

Lens-shaped, biconvex hyperdensity

Tx?

Emergent neurosurgery evacuation

Complications?

Uncal herniation: contralateral homonymous hemianopia, ipsilateral fixed and mid-dilated pupil, contralateral hemiparesis

Subfalcine: compression of anterior cerebral artery branches -> contralateral hemiparesis (lower limbs)

**80 F ground level fall with head strike c/o confusion and motor deficits**

Subdural hematoma

Pathophys?

Rupture of bridging veins -> blood accumulation between dura and arachnoid membranes

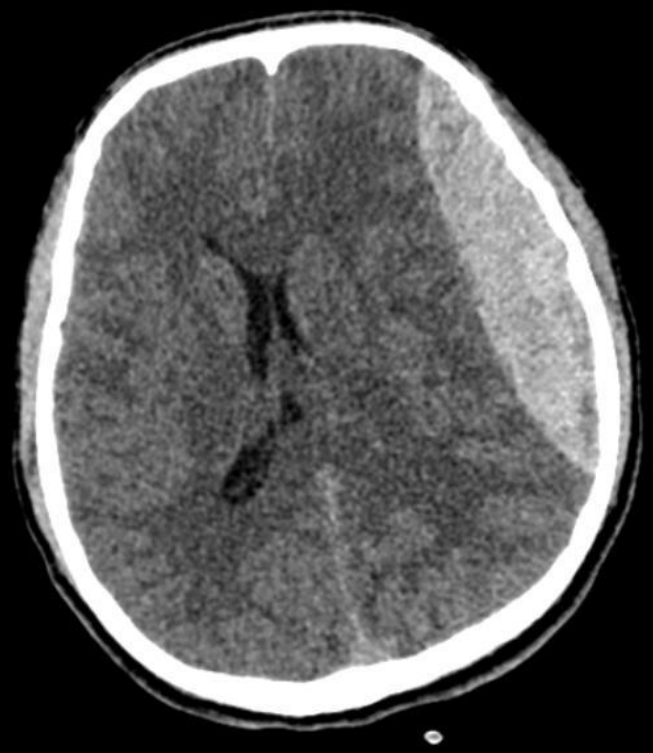
Non-con CT findings?

Crescent shaped, concave hyperdensity (acute)

Tx?

Neurosurgery evacuation

# Epidural



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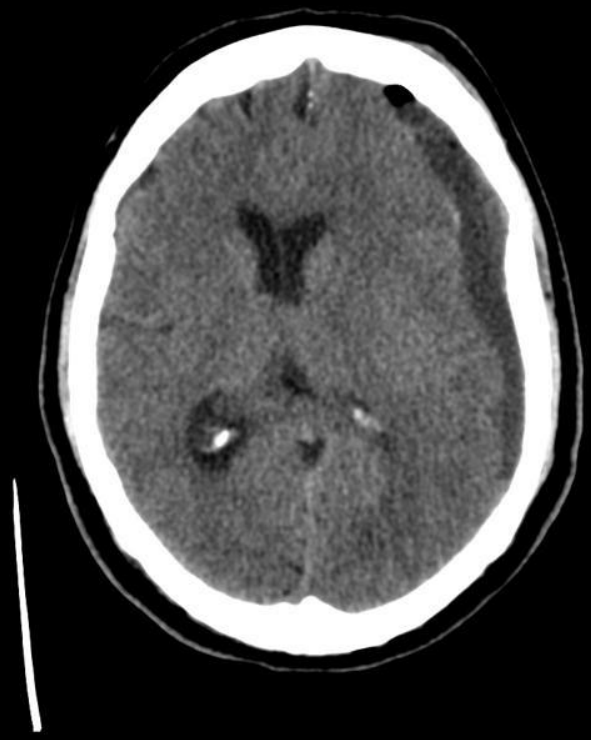
# Subdural



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# Rule of 4s - Brainstem Infarcts

- 4 structures in the midline, beginning with “M”
- 4 structures to the side (lateral), beginning with ”S”
- 4 cranial nerves in the medulla (9-12), 4 in the pons (5-8), 4 above the pons (3 and 4 are in the midbrain)
- 4 motor nuclei that are midline divide equally into 12 -> 3, 4, 6, and 12 (5, 7, 9, and 11 are lateral)

Medial Structures:

- **Motor** (corticospinal tract)
  - Contralateral weakness
- **Medial lemniscus**
  - Contralateral vibration/proprioception loss
- **Medial longitudinal fasciculus**
  - Ipsilateral INO
- **Motor nucleus**
  - Ipsilateral CN loss (3, 4, 6, or 12)

Lateral Structures:

- **Spinocerebellar pathway**
  - Ipsilateral arm/leg ataxia
- **Spinothalamic pathway**
  - Contralateral deficit in pain/temperature sensation
- **Sensory nucleus of CN 5**
  - Ipsilateral alteration of pain/temperature in the face
- **Sympathetic pathway**
  - Ipsilateral ptosis, anhidrosis, and miosis (Horner’s syndrome)

Localizing the lesion

- Which part of the brainstem?
  - Determine which CN(s) are affected
- Medial vs lateral?
  - Determine which tracts are affected

# Localization Examples (1)

Contralateral weakness/sensory loss most pronounced in the upper limbs and lower half of the face?

Middle cerebral artery

Other classic sx if in dominant hemisphere? Non-dominant?

Aphasia (dominant), hemineglect (nondominant)

Contralateral weakness/sensory loss most pronounced in the lower limbs and urinary incontinence?

Anterior cerebral artery

Contralateral homonymous hemianopia with macular sparing?

Posterior cerebral artery

Pure motor stroke?

Posterior limb of the internal capsule (most common)

Pure sensory stroke?

Thalamus (most common)

# Localization Examples (2)

Left arm/leg weakness, no speech difficulty, tongue deviated to right, no vibration sense on the left?

Right medial medullary infarct

Which artery involved?

Anterior spinal artery

Right arm/leg weakness, left eye is deviated down and out, pupil is dilated?

Left medial midbrain infarct

Which artery involved?

Posterior cerebral artery

Left hand ataxia with FNT, loss of sensation to left face, left eye ptosis and miosis, hoarse voice?

Left lateral medullary infarct

Which artery involved?

Posterior cerebellar artery/vertebral artery

# Seizure Disorders Rapid Fire

Common etiologies?

Structural (neoplasm, stroke, developmental abn)

Nonneurologic (infection, metabolic, drugs, trauma)

**Uncontrollable twitching of thumb, fully aware?**

Simple partial seizure

**33 yo with lip smacking, ↓ consciousness w/ confusion?**

Complex partial seizure

Differentiating history to look out for?

Incontinence, tongue biting/oral trauma, postictal state

**8 yo daydreaming/staring with lip smacking and eyelid fluttering for 5-10 seconds at a time?**

Absence seizure

Classic EEG findings?

3 Hz spike and wave discharges

Tx?

Ethosuximide (first line), valproic acid

# Seizure Disorders Rapid Fire

40 M single simple partial seizure 1 minute, 2 mo morning HA and intermittent vomiting. Next step?

Non-con CT head

Pharmacologic therapy for partial & tonic-clonic seizure?

Levetiracetam, phenytoin, carbamazepine, valproic acid (similar efficacy)

**Seizure > 5 minutes or ≥ 2 seizures without return to baseline?**

Status epilepticus

Best initial step?

ABCs (MEDICAL EMERGENCY)

Best initial therapy?

IV lorazepam

If seizures persist?

Consider IV fosphenytoin, IV valproate, or IV levetiracetam

**28 yo M brief tonic-clonic seizures 24 hours after MVC?**

Alcohol withdrawal seizure, treat with IV lorazepam

# Seizure Disorders Rapid Fire

**28 yo M brief tonic-clonic seizures 24 hours after MVC?**

Alcohol withdrawal seizure, treat with IV lorazepam

# Vertigo Rapid Fire

**Severe postural/gait instability, purely vertical or purely torsional nystagmus, focal neurologic signs?**

Central vertigo -> lesions in CNS

Deafness, tinnitus, horizontal torsional nystagmus, absent focal neurologic signs?

Peripheral vertigo -> lesions in inner ear

Episodic vertigo < 1 min triggered by head movements?

BPPV (displaced otoliths from semicircular canals)

Dx and Tx?

Dix-Hallpike and Epley maneuvers

# Vertigo Rapid Fire

## Vertigo, vomiting, hearing loss 1 week after viral infection?

Acute peripheral vestibulopathy (labyrinthitis/vestibular neuritis)

Tx?

Consider corticosteroids, meclizine, antiemetics but typically subsides spontaneously within weeks to months

# Vertigo Rapid Fire

**Episodic vertigo, tinnitus, aural fullness, and hearing loss lasting minutes to hours?**

Meniere's disease

Pathophys?

Increased volume of endolymph (endolymphatic hydrops)

Tx?

Acute: Meclizine/benzodiazepines for spinning, antiemetics for N/V

Chronic: Limit ***salt***, caffeine, nicotine, alcohol intake; betahistine or diuretics for refractory sx

# Rapid onset fever, HA, neck stiffness, AMS, high WBC and knee/hip flexion when the neck is flexed in immunocompromised pt?

Bacterial meningitis

Most common cause in adults? Teens? Neonates?

Strep pneumo, Neisseria meningitidis, GBS

Best next step?

LP for CSF analysis, Gram stain, and culture (before initiation of antibiotics)

If elevated ICP or focal neurologic deficits?

Empiric antibiotics plus CT head before LP

Empiric antibiotic regimen in < 1 mo? Adults?

< 1 mo: Ampicillin + cefotaxime or gentamicin

Adults: IV vancomycin + ceftriaxone or cefotaxime

**> 50 years old/alcohol use disorder/chronic illness/immunocompromised add what?**

Ampicillin

What else is administered to reduce risk of hearing loss?

Dexamethasone (particularly in Strep pneumo meningitis in adults, and Hib meningitis in kids)

**College student with rapid onset fever, neck stiffness, petechial/purpuric rash?**

Meningococcal meningitis

Treatment for close contacts?

Rifampin or ceftriaxone (preferred in pregnancy)

**40 M PMH AIDS with subacute HA, fever, impaired mentation, signs of increased ICP?**

Cryptococcal meningitis

Dx?

CSF pattern from LP, cryptococcal antigen test in CSF (most sensitive/specific), CSF India ink stain

Tx?

Amphotericin B + flucytosine and then extended period of fluconazole

# CSF Profiles

| | Normal | Bacterial | Viral | Fungal/TB |
|---|---|---|---|---|
| **Pressure (cmH20)** | 5-20 | > 30 | Normal or mildly increased | |
| **Appearance** | Normal | Turbid | Clear | Fibrin web |
| **Protein (g/L)** | 0.18-0.45 | > 1 | < 1 | 0.1-0.5 |
| **Glucose (mmol/L)** | 2.5-3.5 | <2.2 | Normal | 1.6-2.5 |
| **Gram stain** | Normal | 60-90% Positive | Normal | |
| **Glucose - CSF:Serum Ratio** | 0.6 | < 0.4 | > 0.6 | < 0.4 |
| **WCC** | < 3 | > 500 | < 1000 | 100-500 |
| **Other** | | 90% PMN | Monocytes<br>10% have >90% PMN<br>30% have >50% PMN | Monocytes |

From Chris Nickson, https://litfl.com/csf-analysis/

**55 M PMH HIV/AIDS with fever, confusion, HA, seizures and multiple ring enhancing lesions?**

Toxoplasmosis

Tx?

Pyrimethamine + sulfadiazine and leucovorin

Prophylaxis?

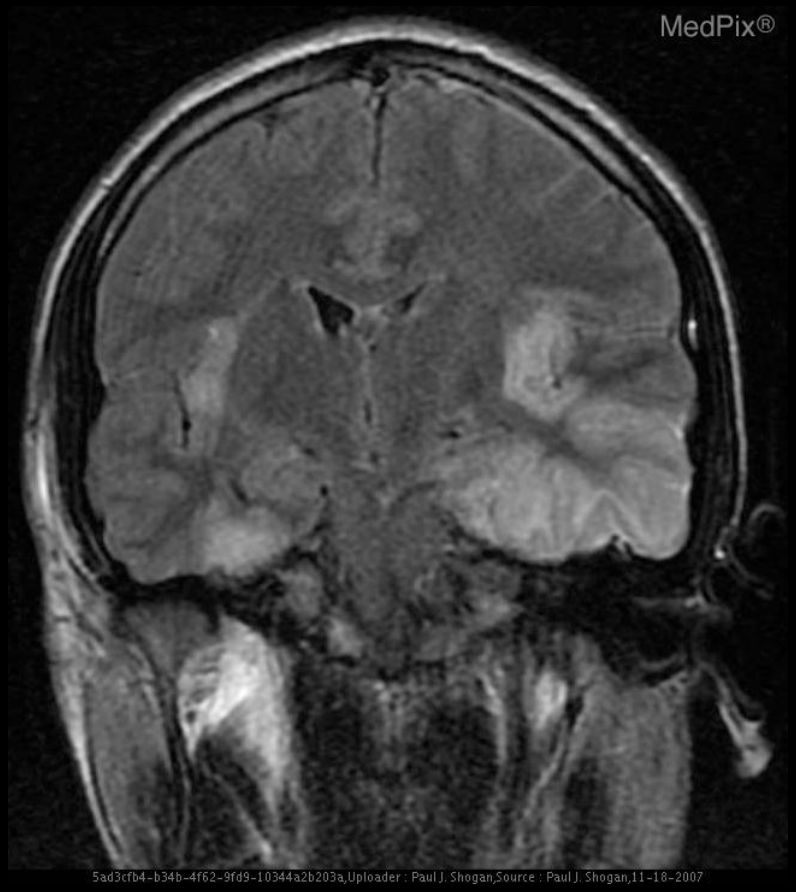
Trimethoprim-sulfamethoxazole if CD4 <100 and positive toxoplasmosis IgG

**22 M with confusion, HA, fever, seizures, RBCs in CSF and provided CT scan (right)?**

HSV encephalitis

Tx?

IV acyclovir




From NIH MedPix. Original Source and Author Paul J. Shogan

**Recent paranasal sinusitis with HA, fever, confusion, FNDs?**

Brain abscess

Most common bacterial causes?

Viridans streptococcus, staph aureus

# 30 F with ptosis, diplopia, bulbar symptoms that worsen at the end of the day, no autonomic dysfunction, initially normal reflexes that worsen with testing?

Myasthenia gravis

Pathophys?

Autoantibodies against nicotinic acetylcholine receptors on the postsynaptic membrane

Best initial *imaging* test?

Chest CT to evaluate for thymoma

Acute exacerbation/myasthenia crisis treatment?

Plasmapheresis or IVIG +/- steroids

Chronic maintenance treatment?

Acetylcholinesterase inhibitors (pyridostigmine)

# 68 M smoker with proximal muscle weakness that *improves* with repetitive nerve stimulation?

Lambert Eaton myasthenic syndrome

Most common association?

Paraneoplastic syndrome -> small cell lung CA

Pathophys?

Autoantibodies against pre-synaptic voltage-gated calcium channels

Best initial *imaging* test?

Chest imaging (CXR, CT) to evaluate for malignancy

Tx?

Treat underlying malignancy

**Descending paralysis in 34 M farmer that eats canned food?**

Foodborne botulism

Tx?

Equine botulinum antitoxin

**9 mo with bulbar palsies, ptosis, hypotonia; what did they eat?**

Honey; treat with IV human botulism immune globulin

# 30 M hx GI infection 2 weeks ago with 5 days of bilateral ascending paralysis with labile BPs?

Guillain-Barre syndrome

Etiology/Pathophys?

URI/GI infections (*Campylobacter jejuni* most common)

Infection triggers formation of autoantibodies against *gangliosides* of peripheral Schwann cells (demyelination)

Dx?

Decreased nerve conduction velocity; albuminocytologic dissociation (CSF protein > 55 but normal WBC)

Monitor for?

Respiratory failure

Tx?

Plasmapheresis and IVIG

# Patient with ascending weakness, normal CSF with *rapid*, ascending paralysis after hiking trip?

Tick-borne paralysis

Treatment?

remove the tick

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# Neurodegenerative Disorders

Major difference from normal aging?

Loss of functional ability (intact ADLs/IADLs)

Dx criteria for major neurocognitive disorder?

Significant decline in ≥ 1 neurocognitive domains (learning/mem, executive func, attention, perceptual-motor)

MMSE < 24/30, functional impairment of ADLs/IADLs

Reversible causes of dementia not to miss?

Hypothyroid (check TSH), vitamin deficiency (B12, B9, B1), meds (benzos, anticholinergics), pseudodementia, chronic subdural/structural (brain MR) , VDRL/HIV screening (depending on stage)

Most common cause of dementia?

Alzheimer disease, age most important risk factor for sporadic

Early clinical features/findings?

**Short-term memory loss** visuospatial deficits, language difficulty, cognitive impairment that gradually declines

Dx?

Clinical features/neuropsychological testing; supportive findings on MRI -> disproportionate atrophy of medial temporal lobe and hydrocephalus ex vacuo

Treatment?

None curative. Acetylcholinesterase inhibitors (rivastigmine, galantamine, donepezil -> mild-mod), memantine (mod-severe)

**75 M PMH HTN, DM, smoking with abrupt worsening right arm weakness and cognitive deficits?**

Vascular dementia

MR findings?

Multiple cortical and lacunar infarcts

Tx?

Treat risk factors for ASCVD

**55 F with disinhibition, socially inappropriate behavior, apathy, and increased cigarette smoking?**

Frontotemporal dementia (behavioral variant is most common)

Dx?

Clinical. MRI -> frontal and/or temporal lobe atrophy

Tx?

Supportive

# Neurodegenerative Disorders

**70 M with ataxic gait, urinary incontinence, and executive dysfunction/decreased attention?**

Normal pressure hydrocephalus

Pathophys?

↓ CSF absorption -> ↑ CSF -> ventricular enlargement -> stretching of corona radiata

Best initial imaging? Findings?

MRI. Ventriculomegaly *without* cortical atrophy/sulcal enlargement

Best confirmatory test?

Improvement of symptoms following LP

Definitive tx?

Ventriculoperitoneal shunt

# Neurodegenerative Disorder

**Rapidly progressive dementia and startle myoclonus?**

Creutzfeldt-Jakob disease

CSF findings? EEG? MRI?

↑ 14-3-3 protein, periodic sharp wave complexes, hyperintensity in the caudate and putamen (“hockey stick sign”)

Tx?

Symptomatic management-poor prognosis

(Unfortunately most patients die within 1 year)

# Neurodegenerative Disorders

**68 M with dementia, fluctuating cognition, flailing arms during sleep, visual hallucinations and later Parkinsonian features?**

Lewy Body dementia

Differentiate LBD from dementia 2/2 Parkinson disease?

Cognitive and motor symptom onset < 1 year apart

Pathologic examination?

Round, eosinophilic neuronal inclusions of alpha-synuclein (Lewy bodies)

Drug sensitivity?

Typical > atypical antipsychotics (can cause severe parkinsonism)

Tx?

Acetylcholinesterase inhibitors for dementia

PD treatment (slower titration) for Parkinsonian features

# 40 M FHx of similar with random, irregular movements, executive dysfunction, and irritability?

Huntington disease

Pathophys?

CAG repeats -> abnormal huntingtin protein -> glutamate excitotoxicity -> loss of GABA/Ach, unbalanced dopamine activity

Classic imaging findings?

Atrophy of the caudate nucleus and putamen, dilation of lateral ventricles

Tx?

Mainly supportive (meds for mood, agitation), no cure

Chorea -> VMAT2 inhibitors (tetrabenazine)

## 62 F with unilateral resting tremor, muscle rigidity, slow shuffling gait, and increasing falls?

Parkinson disease

Pathophys?

↓ dopaminergic/↑ cholinergic activity with loss of dopaminergic neurons in the substantia nigra pars compacta

Tx?

Most effective -> Carbidopa-levodopa

Initial in < 65 yo: nonergot DA receptor agonists (pramipexole, ropinirole)

Tremor predominant: benztropine/trihexyphenidyl

PD patient who develops psychosis?

Trial dose reduction or add quetiapine

Other antidopaminergic drugs to avoid?

Metoclopramide, prochlorperazine, other antipsychotics

**Parkinsonism + dysautonomia (mainly orthostasis)?**

Multisystem atrophy (Shy-Drager syndrome)

**Parkinsonism + vertical gaze palsy (especially downgaze)**

Progressive supranuclear palsy

# 55 M with slowly progressive weakness, increased LUE reflexes, and fasciculations?

Amyotrophic lateral sclerosis

Most common cause of death?

Respiratory failure -> NIPPV for respiratory insufficiency

Any medications delay disease progression?

Riluzole -> decreases glutamate excitotoxicity

## 50 F with irresistible urge to move legs at night, improves with movement?

Restless leg syndrome

Associated with what mineral deficiency?

Iron

Tx?

Gabapentin, pregabalin (first line)

Dopamine agonists (pramipexole, ropinirole)

# 40 M with tremor, parkinsonism, dementia, and jaundice/elevated LFTs?

Wilson disease

Pathophys?

Mutation in intracellular copper transporter (*ATP7B*) leads to accumulation of copper in liver, brain, kidneys, cornea

Tx?

Copper chelation (penicillamine, trientine, zinc)

**65 M with 10 years of bilateral hand tremor that are worse with movement, + FH. No other neurologic symptoms, dx?**

Essential tremor

Classically improves with?

Alcohol

Tx?

Propranolol or primidone

# Neoplasms/Neurocutaneous Disorders

Most common intracranial neoplasm?

Metastatic (70%): primary lung, breast, kidney, GI, melanoma

Most common primary neoplasm in adults? Kids?

Adults: Glioblastoma multiforme, meningiomas

Kids: Astrocytomas, followed by medulloblastomas

Common presenting sx?

Seizures and slowly progressive focal motor deficits

Other sx: HA, visual field deficits, vomiting, etc

# Neoplasms/Neurocutaneous Disorders

**Paralysis of upward gaze, ataxia, HA, emesis, and papilledema?**

Parinaud syndrome

Pathophys?

Lesion in superior colliculus/pretectal area (e.g., pinealoma)

# Neoplasms/Neurocutaneous Disorders

**Patient with flat, hyperpigmented macules, axillary freckling, pigmented iris hamartomas?**

Neurofibromatosis 1

Screen children under 6 for (optho)?

Optic glioma

20 F with tinnitus, hearing loss, and vertigo?

Neurofibromatosis 2

Next step?

MRI brain and spine w/ contrast -> bilateral vestibular schwannomas

# Neoplasms/Neurocutaneous Disorders

**Child with seizures, hypopigmented lesions on trunk/extremities, red nodules on nose/cheeks, and learning disability?**

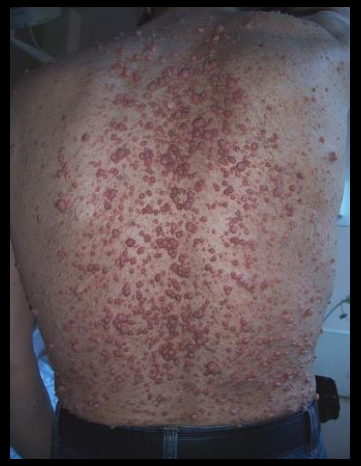
Tuberous sclerosis

Associated CNS neoplasm?

Subependymal giant cell astrocytoma (5-15% of patients)

## Neurofibromatosis Type 1 (NF1)

- Cutaneous neurofibromas, gliomas/meningioma, café au lait, Lisch nodules, scoliosis
- Pheochromocytoma
- Mutation in NF1 suppressor gene
  - Chromosome 17, Autosomal dominant



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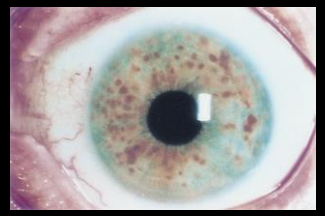
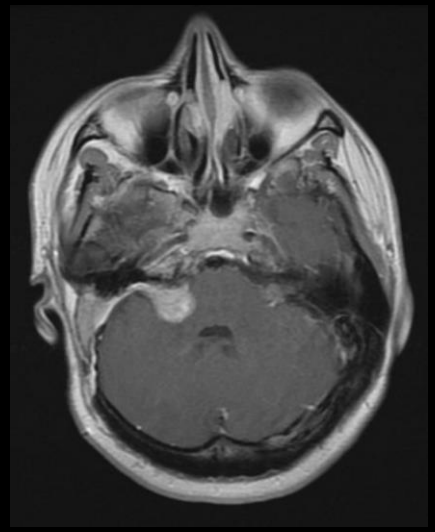


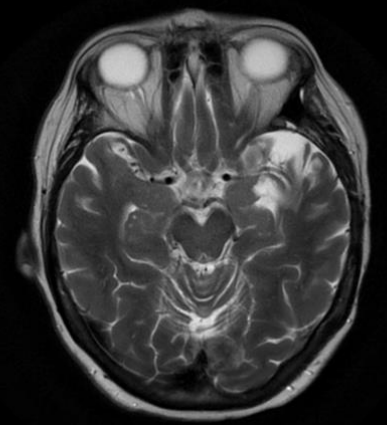
American Academy of Ophthalmology

## Neurofibromatosis Type 2 (NF2)

- Presents with bilateral schwannomas, cataracts, multiple meningiomas, ependymoma.
  - Café au lait spots, neurofibroma less common
  - Schwannoma at cerebellopontine angle (VI and VIII)-hearing loss-get MRI
- Mutation in NF2 suppressor gene
  - Chromosome 22, autosomal dominant



Acoustic schwannoma of cerebellopontine angle
Case courtesy of Frank Gaillard, Radiopaedia.org, rID: 2575

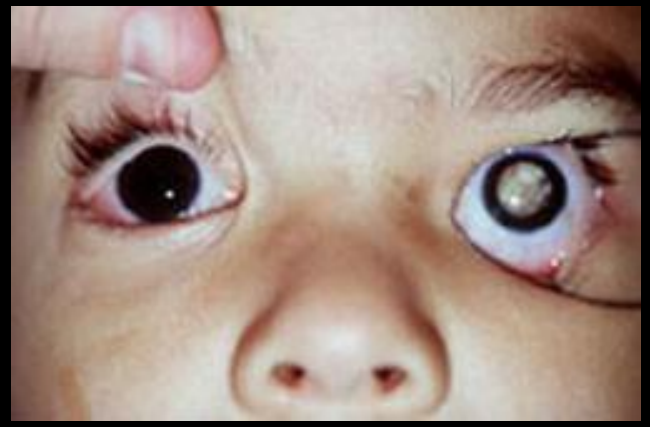


https://radiopaedia.org/cases/neurofibromatosis-type-2-with-multiple-meningiomas-1?lang=us

Case courtesy of Bruno Di Muzio, Radiopaedia.org, rID: 38271

## Retinoblastoma

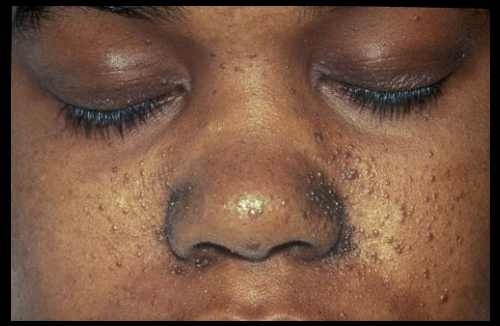
- Presents with **white reflex (leukocoria)** in infants
  - Part of standard newborn exam
  - Sporadic vs hereditary
- Increased risk for osteosarcoma
- Mutation in Rb1 gene
  - Normally inhibits E2F to block G1 to S



American Association for Pediatric Ophthalmology and Strabismus

## Tuberous Sclerosis

- Hamartomas
- Initially presents with seizures
  - subependymal nodules (hamartomas of ventricles)
- Angiofibroma, Ash leaf spot, Cardiac Rhabdomyoma, Renal Angiomyolipoma, Shagreen patches
- TSC1 or TSC2 mutation (hamartin, tuberin)
  - Autosomal dominant
  - Often variable expression



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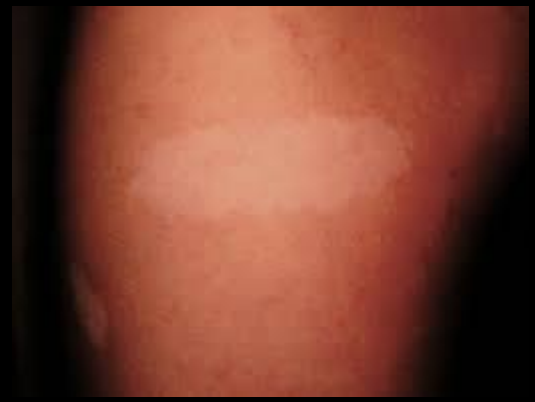
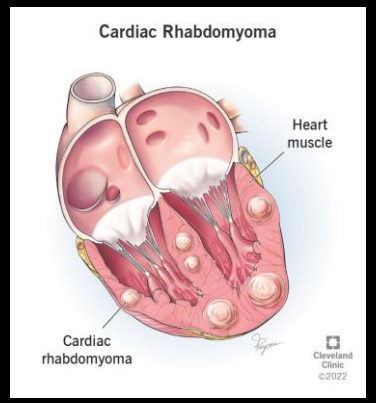


DermNet




Cleveland Clinc

# 10 F with seizures, intellectual disability, and a red/purple patch on the right side of the face?

Sturge-Weber syndrome (port wine stain)

Classic MRI findings?

Leptomeningeal angioma (“tram track appearance”)

**Syndrome associated with hemangioblastomas?**

Von Hipple-Lindau

- Common in retina, cerebellum, brainstem and/or spine

Other HY associations?

Bilateral renal cell carcinoma and pheochromocytoma


https://dermnetnz.org/topics/capillary-vascular-malformation

## 55 M with encephalopathy, ataxic gait, and deficits with eye movement?

Wernicke encephalopathy

Tx?

Reversible: thiamine and then glucose

## 60 M PMH alcohol use disorder with new onset confabulations and anterograde amnesia?

Korsakoff syndrome

Tx?

Irreversible: Wernicke that was treated inadequately or too late

# Thank you for watching!

By Evan, Mike

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